

Patent claims

1. Nucleic acid, which includes

- a) the nucleotide sequence shown in Fig.1 or a protein-coding segment thereof,
 b) one of the sequence from a) within the context of the degeneration of the genetic code corresponding nucleotide sequence,
 c) one with the sequences from a) and/or b) under stringent conditions hybridizing nucleotide sequence, except for the EST sequences:

10 AA165403, AA455594, AA314472, N34087, AA452340, AA182700, N41615, AA470049, AI751597, AA463289, AA132459, W31487, R56355, H58271, H16122, W77956, AA193332, AA323923, AA370209, AA296758, W72757, AA093971, AA385544, AA386175, AA165402, AW083713, H42806, AA093977, AI161152, AA370011, AI671702, R71215, AA885343, T79297, AI814869, R81567, AI082713, N29615, AW087726, AW075710, AI952608, AI818073, AI784445, AI432812, AI375568, AI372904, AI364106, AI143379, AA993074, AA953985, AA862385, AA761084, AA576229, AA569223, AA463198, AA452117, AA416877, AA074872, W16851, W04568, N40176, AW068354, AA857004, H58663, H15819, AW264944, AI923965, AI692214, AI475321, AI435987, AA961068, AA206059, AI469161, T84789, AA507257, AA707515, AA132458, AA179262, T79211, W31505, N25699, T99574, T99363, AI751598, AA713668, T91119, AW105515, AA370208, AI422128, R81568, AI038899, AI971847, AI540650, AI826106, AA885960, R56263, AA825431, T99147, D31503 and AF049564, or

- d) a complementary sequence to the sequences of a) and/or b).

2. Nucleic acid according to claim 1, which includes a protein-coding segment comprising of preferably at least 30 nucleotides of the nucleotide sequence shown in Fig. 1.

3. Nucleic acid, which shows a homology of more than 65% with the nucleotide sequence according to claim 1 or a segment thereof.

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4. Modified nucleic acid or nucleic acid analogue, which includes a nucleotide sequence according to one of the claims 1 to 3.
5. Recombinant vector, which includes at least one copy of a nucleic acid according to one of the claims 1 to 3 or a section thereof.
6. Recombinant vector according to claim 5, which enables the expression of the nucleic acid in a suitable host cell.
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- 10 7. With a nucleic acid according to one of the claims 1 to 3 or a vector according to claim 5 or 6 transformed cell, a corresponding non-human transgenic organism or animal models, which stably produce (knock-in) the product of the nucleic acid according to one of the claims 1 to 3 or whose corresponding natural gene was destroyed deliberately (knock-out).
- 15 8. Polypeptide or a salt thereof, which is coded by a nucleic acid according to one of the claims 1 to 3.
9. Polypeptide according to claim 8, which exhibits
- a) the amino acid sequence shown in Fig. 2 or
- 20 b) a homology of more than 60% with the amino acid sequence shown in Fig. 2 or a salt thereof.
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- 25 10. Fragment of the polypeptide according to claims 8 or 9 with at least 100 amino acids or salts thereof.
11. Modified polypeptide, which includes an amino acid sequence according to claims 8 or 9.
12. Methods for the synthesis of the polypeptide according to claim 8 or 9, which includes the cultivation of cells according to claim 7 as well as the isolation of the polypeptide according to claim 8 or 9.
- 30 13. Use of a polypeptide according to claim 8 or 9 or of fragments of this polypeptide as an immunogen for the production of antibodies.

14. Antibodies against a polypeptide according to claim 8 or 9.
15. Method for the identification of effectors of a protein according to claim 8 or 9,
5 with the help of which various potential effector substances can be tested on cells,
which express the protein.
16. Pharmaceutical composition, which includes as active component
- 10 a) a nucleic acid according to one of the claims 1 to 4,
b) a vector according to claim 5 or 6,
c) a cell according to claim 7,
d) a polypeptide according to claim 8, 9, 10 or 11,
e) an antibody according to claim 14
and which contains the pharmaceutically usual carrier, auxiliary and/or additive
15 substances.
17. Use of a composition according to claim 16 for diagnosis of diseases, which are
associated with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis
and/or tumor progression, or a predisposition to such diseases.
- 20 18. Use of a pharmaceutical composition for diagnosis of diseases which are associated
with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or
tumor progression, or a predisposition to such diseases, which contains as an active
component
- 25 a) an EST sequence according to claim 1c,
b) a recombinant vector which includes at least one copy of the EST sequences
mentioned above,
c) a recombinant vector according to b) which enables the expression of the
nucleic acid in a suitable host cell,
- 30 d) a cell according to claim 7, whereas the nucleic acid consists of one of the EST
sequences mentioned above,
e) a polypeptide being coded by one of the EST sequences mentioned above or a
salt thereof or,

- f) a polypeptide according to e) which exhibits the amino acid sequence shown in Fig.2 or a homology of more than 60% with the amino acid sequence shown in Fig.2 or a salt thereof,
- g) a fragment of the polypeptide according to e) or f) with at least 100 amino acids or a salt thereof,
- h) a modified polypeptide which includes an amino acid sequence according to e) or f),
- i) an antibody against a polypeptide according to e) or f) and which contains pharmaceutically usual carrier, auxiliary and/or additive substances.
19. Use of a composition according to claim 16 for the therapy or prevention of diseases, which are associated with DNA repair defects, cell cycle disorders, cytopenia, tumor genesis and/or tumor progression.
20. Use of a pharmaceutical composition according to claim 18 for the therapy or prevention of diseases, which are associated with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or tumor progression.
21. Use of a composition according to claim 16 for a gene therapy of diseases, which are associated with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or tumor progression.
22. Use of a pharmaceutical composition according to claim 18 for gene therapy of diseases which are associated with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or tumor progression.
23. Methods for diagnosing diseases, which are associated with DNA repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or tumor progression or a predisposition to such diseases, during which a patient or a sample from the patient is brought in contact with a composition according to claim 16 and the nucleotide sequence and/or the expression of a nucleic acid according to claim 1 is determined.
24. Methods for the therapy or prevention of diseases, which are associated with DNA

repair defects, cell cycle disorders, cytopenia, tumorigenesis and/or tumor progression, during which a patient is administered a composition according to claim 16, which contains the active components in an amount effective against the disease.

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